LETTERS TO THE EDITOR

Byler’s syndrome

Editor,—The report of Byler’s syndrome with raised sweat electrolytes in an Irish traveller kindred1 interests us, as we have observed raised sweat electrolytes in two members of the original Byler kindred who have Byler’s disease. Neither has cystic fibrosis; both underwent liver transplantation in their second decade and subsequently developed pancreatic disease. One has had recurrent pancreatitis and the other has a fibrotic operated pancreatic disease. One has had recurrent pancreatitis or evidence of pancreatic dysfunction. As these children likely will need further evaluation and/or transplantation in the coming years we will have the opportunity to undertake further studies including analysis of biliary bile acid content and examination of biopsy samples for the presence of ‘Byler bile’.

We are aware of the report of Kinsely et al describing pancreatic disease in members of the original Amish kindred with Byler’s disease.2 Although we have not observed pancreatitis or evidence of pancreatic dysfunction in the Irish traveller family with Byler’s syndrome, one of us (BB) has encountered a child with progressive familial intrahepatic cholestasis and chronic pancreatitis at another institution (patient of E Roberts and R Superina, Hospital For Sick Children, Toronto). Whether this child has a mutation at 18q21-q22 is currently being evaluated.

The presence of raised sweat electrolytes and pancreatitis in a subset of these children with Byler’s disease/syndrome is certainly intriguing and raises interesting questions about the function of the mutated allele at 18q21-22. Ongoing genetic studies of members of the original kindred and unrelated families such as this Irish family should soon provide answers to these questions.


Figure 1 Pedigree of family with Byler disease-like progressive familial intrahepatic cholestasis described by Lloyd-Still.1

Figure 2 Transmission electron micrograph of coarsely granular bile, characteristic of bile from children with Byler’s disease,1 within canaliculus of liver obtained at hepatectomy in affected boy (III.2); 4% paraformaldehyde/0.5% glutaraldehyde in Sorenson’s phosphate buffer, pH 7.3; OvO4/uranyl acetate/lead citrate (original magnification × 18 000).
Intestinal neuronal dysplasia associated with cystic fibrosis

**EDITOR,—**The association between cystic fibrosis and intestinal neuronal dysplasia (IND) has been rarely described. We report a case of full thickness, biopsy proved, IND type B of the ileum and colon associated with cystic fibrosis. The boy was born at full term to non-consanguineous parents. Because of obstructive symptoms, several resections were performed: 20 cm of distal ileum after birth; distal ileum and part of ascending colon at the age of 18 days; ileum, part of jejunum, and colon at the age of 2 months. A series of radiographs of the upper gastrointestinal tract series showed a normal duodenum at 16 months and no dilatations of the remaining intestinal tract. Contrast appeared in the rectum after 90 minutes. By histology, the proximal ileal tract had 6.25 neurons/mm of myenteric plexus, according to Smith's method (normal values: 2–4), the ascending colon 16.0 neurons/mm, and the transverse colon 8.0 neurons/mm. Acetylcholinesterase staining showed an increase of number of submucosal ganglia, neuronal heterotopy, and increase of positive fibres in circular muscular layer and lamina propria. NADPH-DH showed an increased number of neurons in myenteric and submucosal plexuses. The study with neurofilaments (NF65, NN18) showed a normal maturity of neurons. The results of two sweat tests were abnormal. An homozygosity for the delta F508 mutation was demonstrated and both parents were carriers of the allele. It is possible that there is an NID-B determining gene linked to the cystic fibrosis transmembrane conductance regulator locus for cystic fibrosis, localised on chromosome 7q.

We suggest that intestinal neuronal dysplasia should be considered as an underestimated, underlying cause in patients with cystic fibrosis having functional small bowel dysmotility and obstruction leading to emergencies, such as meconium ileus in neonates or meconium ileus equivalent in children and adults.

**A TOTZZI**
**G ASCIONE**
**M L CARPENTIERI**
**A STAiano**

*Departments of Paediatrics and Division of Paediatric Surgery*,
*University Federico II, Via S Panzani 5, 80131 Naples, Italy*

Situs inversus and left sided pyloric tumours

**EDITOR,—**The case report by Harrington et al reminded me of the procedure to examine for a pyloric tumour taught by Dr M J Simpkins, based on an identical case he had seen decades previously. Namely, define the apex beat before examining the abdomen. This combination will occur again, just like the case of ‘The glass eye’.

**RICHARD SPORIK**
*Institute of Respiratory Medicine, University of Sydney, NSW 2006, Australia*